

Indications for prenatal cytogenetic testing

Currently our prenatal testing policy is dependent on the basis of funding for the service provided to a particular User group. Funding for the majority of patients is provided by the Specialist Genetics Commissioners for the London Region. In addition we provide a service to patients from some Hospital Trusts with whom we have a Service Level Agreement. This distinction is important because the service we provide to these two groups is different.

Specialist Genetics Commissioned Services

Rapid prenatal aneuploidy testing and sex determination by QF-PCR

- Rapid testing for trisomy 13, 18 and 21 by QF-PCR is performed on all amniotic fluid and chorionic villus samples unless a patient chooses to opt out. This test is not equivalent to a full chromosome analysis but will detect the common clinically significant trisomies.
- Rapid chromosomal sexing by QF-PCR (by X and Y copy number), is not performed routinely, but can be undertaken on amniotic fluid and chorionic villus samples where this is considered to be of clinical value, for example where there is a clinical indication of risk for Turner syndrome or where there is a risk of sex linked single gene disorder. This is not equivalent to a full chromosome analysis and karyotyping may be required to confirm the result.

Prenatal Karyotyping

From May 2007, the Genetics Commissioners will only fund prenatal karyotyping for pregnant women falling into one the following categories:

- Abnormal ultrasound scan findings which are indicative of chromosome abnormality:
 1. Ultrasound detection of structural abnormality including ventriculomegaly, echogenic bowel, renal pelvic dilatation and IUGR – i.e. small measurements compared to dating scan.
 2. nuchal translucency > 3mm before 14 weeks gestation, or a nuchal fold measuring 6mm or greater between 14 and 20 weeks gestation.
- A history of chromosome abnormality indicates an increased risk for future pregnancies. In particular:
 - The woman or her partner is a carrier of a chromosomal rearrangement
 - A previous pregnancy was chromosomally abnormal (excluding common trisomies detectable by rapid aneuploidy testing)
 - There is a family history of chromosome abnormality (karyotyping of the woman or her partner should be undertaken first in order to establish whether prenatal karyotyping is indicated)

The Genetics Commissioners will no longer fund prenatal karyotyping for pregnant women where:

- Screening for Down syndrome indicates that there is an increased risk of carrying a fetus with Down syndrome. Women in this group are now offered rapid prenatal aneuploidy testing by QF-PCR for trisomy 13, 18 and 21 (Down syndrome) only.

or

- The primary reason for referral is a monogenic genetic disorder (e.g. Cystic Fibrosis).

Women who fall into either of these groups may opt to have prenatal karyotyping on a private patient basis (see below).

If a patient falls outside these referral categories, please contact the Prenatal Section Head, Thomas Spencer to discuss the test required before sending a sample. Maternal anxiety is not an indication for prenatal karyotyping other than in exceptional circumstances where the referring clinician considers it to be important for the management of the pregnancy.

Services for patients referred from Hospital Trusts with whom we have a Service Level Agreement

Patients from Hospital Trusts with whom we have a Service Level Agreement can be offered both Karyotyping and rapid prenatal aneuploidy testing by QF-PCR for trisomy 13, 18 and 21 (Down syndrome) or rapid prenatal aneuploidy testing alone.

Please note that:

- Rapid testing for trisomy 13, 18 and 21 by QF-PCR is performed on all amniotic fluid and chorionic villus samples unless a patient chooses to opt out. This test is not equivalent to a full chromosome analysis but will detect the common clinically significant trisomies.
- Rapid chromosomal sexing by QF-PCR (by X and Y copy number), is not performed routinely, but can be undertaken on amniotic fluid and chorionic villus samples where this is considered to be of clinical value, for example where there is a clinical indication of risk for Turner syndrome or where there is a risk of sex linked single gene disorder. This is not equivalent to a full chromosome analysis and karyotyping may be required to confirm the result.

Optional privately funded karyotyping

Karyotyping (full chromosome analysis) is available on request for all prenatal samples from patients where this test would not normally be offered. For details of costs and method of payment, please contact the Laboratory Administrator, Michael Tinsley.

For further details regarding all the above policies please contact the Prenatal Section Head, Thomas Spencer.